Macroscopic and mesoscopic cortical organization in congenital visual pathway abnormalities

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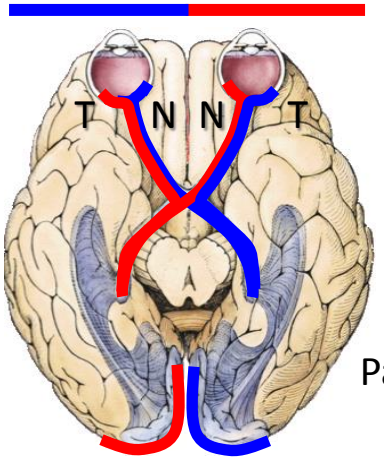
Overview

- **Congenital visual pathway abnormalities**
  - Definition & types
  - Retinotopic organization *(macroscopic level)*
  - Columnar organization *(mesoscopic level)*
  - Scope of plasticity and stability

Introduction

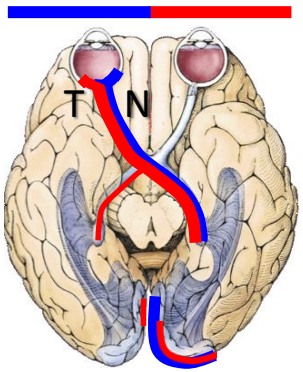
- **Optic nerve projections in normal brain**
  - Contralateral projection of nasal retinal fibers
  - Ipsilateral projection of temporal retinal fibers
  - Representation of each hemifield on contralateral hemisphere



Partial decussation of retinal fibers at optic chiasm

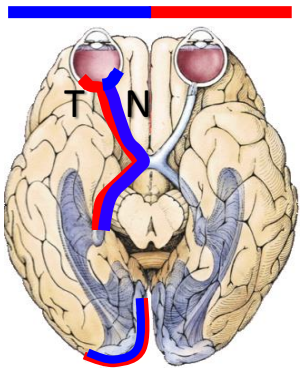
Introduction

- **Optic nerve projections in congenital visual pathway abnormalities**
  - Enhanced crossing of temporal fibers as in albinism & **FHONDA**
  - Reduced crossing of nasal fibers as in achiasma
  - Retinotopic representation of both hemifields on each hemisphere



Albinism

Approx. 1/17000



Achiasma

Approx. 50 published cases

Hoffmann & Dumoulin. Trends in Neurosciences (2015)

Introduction – Characteristics

Albinism

- Hypopigmentation of eye, skin & hair
- Foveal hypoplasia
- Reduced visual acuity
- Nystagmus & strabismus
- Absent Stereo vision
- Overlapping retinotopic maps
- Intact visual perception

Achiasma

- Absence / hypoplasia of optic chiasm
- Normal fovea
- Reduced visual acuity
- Nystagmus & strabismus
- Absent Stereo vision
- Overlapping retinotopic maps
- Intact visual perception

Introduction – Characteristics

FHONDA

A novel congenital visual pathway disorder

- Foveal Hypoplasia
- Optic Nerve Decussation defects
- Anterior segment dysgenesis
- Prevalence of 1 in 1000000
- Absence of pigmentation deficits ► **NO albinism**

Al-Araimi et al., Molecular vision (2013)

Poulter et al., American Journal of Human Genetics (2013)

Research Question

How are visual field maps organized in FHONDA?

Do they follow the observed pattern in albinism and achiasma?

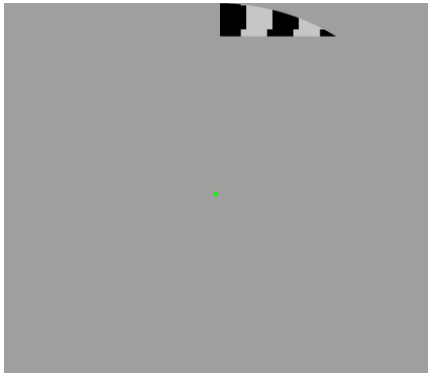
Methods

- **Participants**

  FHONDA = 2, Controls = 2

- **Visual stimuli**

  Monocular stimulation with drifting bar apertures under right & left hemifield conditions



Methods

- **fMRI acquisition**

  Achieva Philips 7 T scanner, multi-slice EPI sequence

  - T1 image: Voxel size = 0.8 mm^3 isotropic
  - T2* images: Voxel size = 2 mm^3 isotropic, TR = 1500 ms & 168 volumes

- **Data analysis**

  - FSL

    Preprocessing of T2* images & segmentation of T1 image

  - MrVista

    Prediction of BOLD response based on population receptive field (pRF) modeling

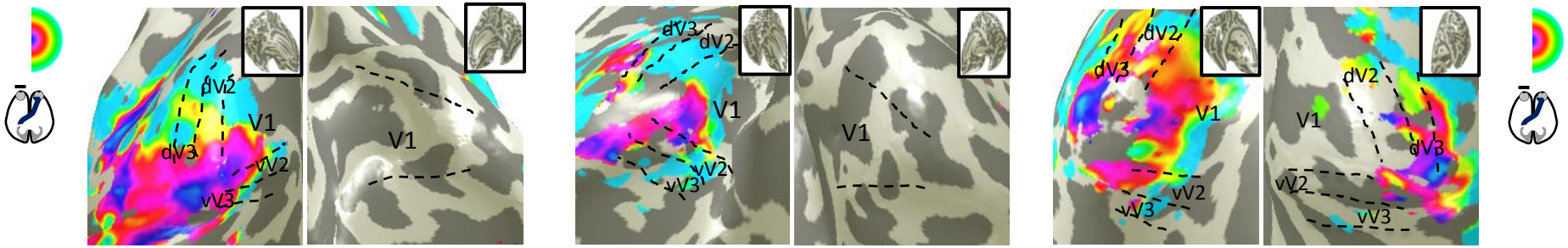
    Dumoulin & Wandell. NeuroImage (2008)

Results – Hemifield maps

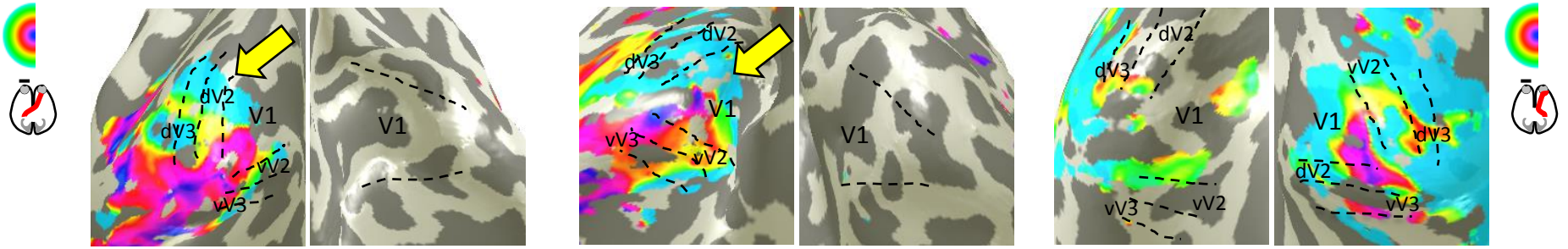
- **Right hemifield mapping**

FHONDA | Control

F1 | F2 | C1



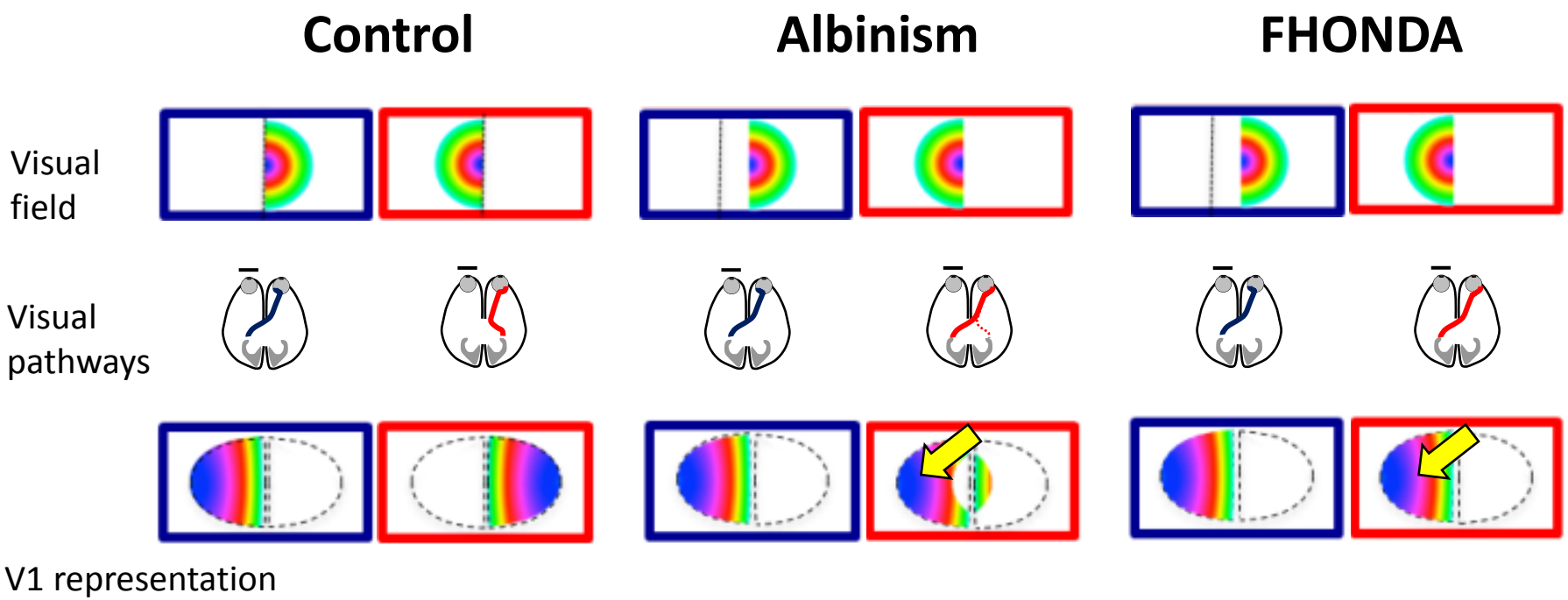
- **Left hemifield mapping**



Results

Similar & overlaid representation of opposing hemifields confined to the contralateral hemisphere in FHONDA

Summary



- Superimposed retinotopic maps of opposing hemifields in FHONDA
- Identical mechanisms shaping the organization of the visual cortex
- Larger extent of misrouting in FHONDA than albinism

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Altered organization of the visual cortex in FHONDA syndrome

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